GENETICS

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The Father of Genetics

Gregor Mendel was born in 1822 in Austria. As a monk he gardened a great deal. He observed that traits from parents seemed to be passed from generation to generation. He studied the inheritance traits in pea plants - discovering the basic laws of inheritance. During his experiment he cultivated over 28,000 pea plants and came up with two generalizations known as Mendel's Law of Inheritance. He is known as the Father of Genetics.

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Mendel's Observations

Mendel kept detailed records of several pea characteristics for generations:
- Height
- Flower color
- Pod color
- Shape of peas and pods
- Flower position

If he had tall plants in one section of the garden, they always produced tall offspring.

Similarly if he had short plants in another section the same would occur (short offspring)

He called these plants Pure Lines
- Tall x Tall = Tall
- Short x Short = Short

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Mendel's Experiment

- Mendel decided to cross (breed) his pure lines

Pure Line Tall x Pure Line Short

- The result was all Tall plants

- He decided to cross these plants with each other
  - this resulted in both tall and short plants
  - the ratio was exactly 3 to 1 (Tall to short)

Parents	Tall (Pure Line) x Short (Pure Line)
First Generation (F1)	All Tall Plants
Second Generation (F2)	Tall & Short Plants 3 to 1 ratio (75% Tall & 25% Short)

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Mendel's Four Conclusions

1) All characteristics are controlled by factors inherited from your parents.

2) There are two factors for every trait (characteristic)
   - Examples of traits include hair color, skin tone, nose length

3) One factor can dominate another ('Mask' the other)

4) These factors separate from each other during sex cell formation (meiosis)

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Modernizing Mendel's Conclusions

- We now call these factors Alleles

- Alleles can be Dominant or Recessive

- We represent Alleles with letters:
  - Capitals = Dominant Alleles
  - Lower Case = Recessive Alleles
  - Dominate alleles are always written first

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Dominant vs. Recessive Traits

- Brown Eyes
- Brown Eyes
- Dark Hair
- Attached Earlobe
- Baldness
- Normal Hairline
- Freckles
- Broad Lips
- Albinism

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Genotype - the combination of alleles you have.

Phenotype - is the expressed traits

Homozygous - 2 alleles that are the same

Heterozygous - 2 alleles that are different

Genotype	Phenotype
TT	Tall Plant
Tt	Tall Plant
tt	Short Plant
BB	Brown Eyes
Bb	Brown Eyes
bb	Blue Eyes

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Types of Genotypes

Genotypes can also be expressed in words

Genotype in Letters	Genotype in Words	Phenotype
TT	Homozygous Dominant	Tall Plant
Tt	Heterozygous	Tall Plant
tt	Homozygous Recessive	Short Plant

'Homo' - Latin prefix for 'the same'

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	B	b
b		
b		

Drag each colored hamster or pea into the correct cell of the Punnett Square.

	R	R
r		
r		

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Mendel's Punnett Square

Mendel started his experiments crossing a pure line of tall plants (TT) with a pure line of short plants (tt).

Parents (TT x tt) Cross	T	T
t		
t		

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Mendel then took those hybrids (Tt) and crossed them with each other.

Parents (Tt x Tt) Cross	T	t
T		
t		

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Sample Problem 1

- Widow's peak is dominant to straight hairline.

- A homozygous male with widow's peak reproduces with a homozygous woman with a straight hairline.

- What hair line will their children (F1) have?

Hairline Cross	W	W
w		
w		

- What genotype in words can we expect from a homozygous dominate cross with a homozygous recessive?

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Sample #1 Continued

- Now if an offspring of that cross reproduces with another of the same genotype, what hairlines can we expect for their children?

- What genotype(s) (in words) can we expect from a cross such as this?

- What ratio of phenotype?

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Our Findings

- Homozygous dominant crossed with homozygous recessive yields heterozygous offspring (100%)

- A heterozygous crossed with heterozygous yields three options:
    - Homozygous dominant
    - Heterozygous
    - Homozygous Recessive

- The ratio of those options are:
    - a genotype ratio of 1:2:1 (HD:H:HR
    - a phenotype ratio of 3:1 (dominant ; recessive)

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Sample Problem #2

- Attached earlobes are recessive to un-attached (hanging) lobes

- A homozygous dominant man (for the trait) mates with a heterozygous woman (for same trait)

- What are the chances of their offspring having attached earlobes?

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Vocabulary-erase to reveal the answer

The passing of traits from parents to offspring is known as ____________

______are different forms of characteristics.

The science of heredity is known as________.

The process of the joining of an egg and sperm cell is __________.

_________organisms are offsprings of many generations that have the same traits.

Factors that control traits are known as______.

_______are different forms of a gene.

_______________are traits always expressed.

________________are those traits masked by dominant traits.

________are organisms that have two different alleles for a trait.

Word bank
heredity, hybrids, alleles, genetics, genes, recessive alleles, fertilization, purebred, dominant alleles

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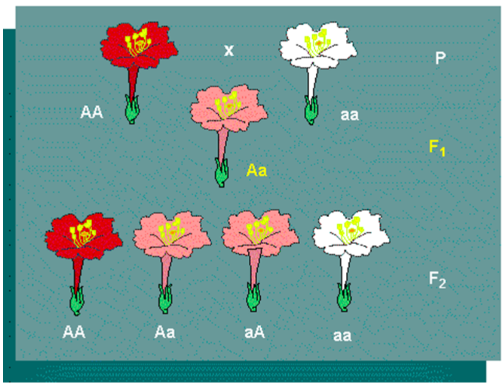
Variations in Dominance

- All of the 8 traits Mendel looked at had two alleles (dominant & recessive)

- Another form is incomplete dominance
  - this is where neither of the alleles can considered dominant and recessive

- The phenotype is a mixing of the two different alleles. Ex. flowers



- Red flowers crossed with white flowers produce pink flowers.

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Incomplete Dominance

- In order to represent these alleles we need to make a slight change ... let's talk about this in reference to flower color.

- Since we're talking about color we'll use the letter C. For each color we add the capital subscript to the allele.

- Capital superscript designations always indicate Incomplete Dominance
    - Therefore:
        - C^R = Red Allele
        - C^W = White Allele

- What the genotype means:
    - C^RC^R = Red flower
    - C^RC^W = Pink flower
    - C^WC^W = White flower
    - C^WC^R = Pink flower

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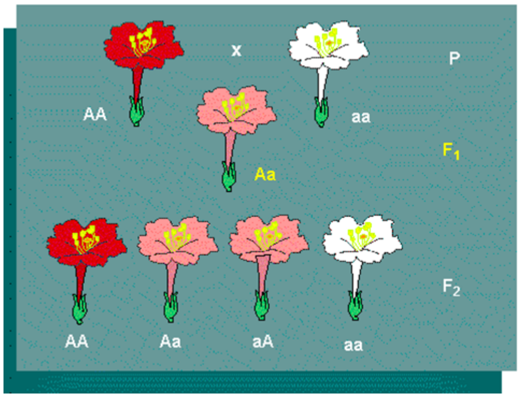
Now cross a red and white flower in the first punnett square then cross two pink flowers in the second punnett square.



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Codominance of Flowers

- With codominance neither allele is dominant; they both show through.

- In codominance a plant with red flowers mating with a plant with white flowers will produce offspring with both red and white flowers on the same plant.

- To represent codominance we must write the alleles differently. Using lower case letters.
    - C^rC^r = Red flowers
    - C^wC^w = White flowers
    - C^rC^w = Both red and white flowers

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Sample Problem

- Let's cross homozygous red plant with a heterozygous one.

- What are the chances of the offspring having red and white flowers?

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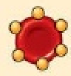
Codominance

- Neither allele is dominant and both alleles are expressed in heterozygous individuals
- Example ABO blood types

Table 14.2 Determination of ABO Blood Group by Multiple Alleles

Genotype	Phenotype (Blood Group)	Red Blood Cells
I^AI^A or I^Ai	A	
I^BI^B or I^Bi	B	
I^AI^B	AB	
ii	O	

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Multiple Alleles

- Some traits are controlled by multiple alleles
(two alleles = 1 gene)

- With multiple alleles there are more than two options for alleles. For example blood type.
  - I^AI^A or I^Ai represent Type A blood
  - I^BI^B or I^Bi represent Type B blood
  - I^AI^B represent Type AB blood
  - ii represent Type O blood

(Type A and Type B blood are codominant)

- Type A, B, and AB blood are all dominant over type O blood

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- What would be the genotype options for a cross between a person with type A blood and a person with type B blood?

- Other examples of multiple alleles include hair color, corn, and feather color (some species).

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A Multiple Allele Cross

Cross fruit fly eye color alleles E^1 and E^3

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Monohybrids & Dihybrids

Monohybrid Crosses - crosses that are looking at <u>one set</u> of alleles. (Everything we have done so far)

	B	b
B	BB	Bb
b	Bb	bb

Dihybrid Crosses - crosses that are looking at <u>two sets</u> of alleles

	RY	Ry	rY	ry
RY	RRYY	RRYy	RrYY	RrYy
Ry	RRYy	RRyy	RrYy	Rryy
rY	RrYY	RrYy	rrYY	rrYy
ry	RrYy	Rryy	rrYy	rryy

The colors in this example are representing different phenotypes

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Dihybrid Example

- In pea plants tall is dominant to short and purple flowers are dominant to white. So if a pea plant is heterozygous for height and flower color what is it's genotype?

TtPp

During the creation of sex cells (meiosis) there are four combinations of alleles the gametes (sex cells) could hold:

TP	dominant and dominant
Tp	dominant and recessive
tP	recessive and dominant
tp	recessive and recessive

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If a pea plant that is heterozygous for both traits reproduced with a plant having the same genotype, what might the phenotype of their offspring be?

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Dihybrid Cross Summary

If we look at the combinations of phenotypes possible from a dihybrid cross between two heterozygous organisms for two traits we find a ratio of: 9:3:3:1

9 = Tall plants with purple flowers
3 = Tall plants with white flowers
3 = Short plants with purple flowers
1 = Short plants with white flowers

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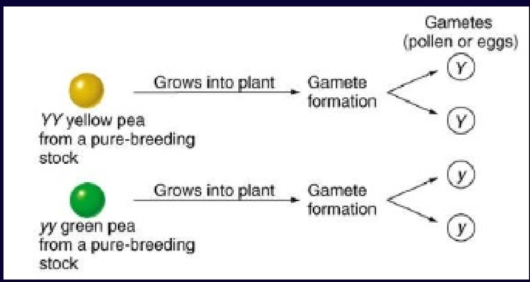
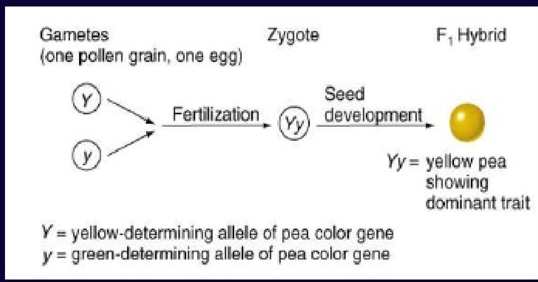
Law of Segregation

- Mechanism of gene transmission

Gametogenesis: alleles segregate



Fertilization: alleles unite



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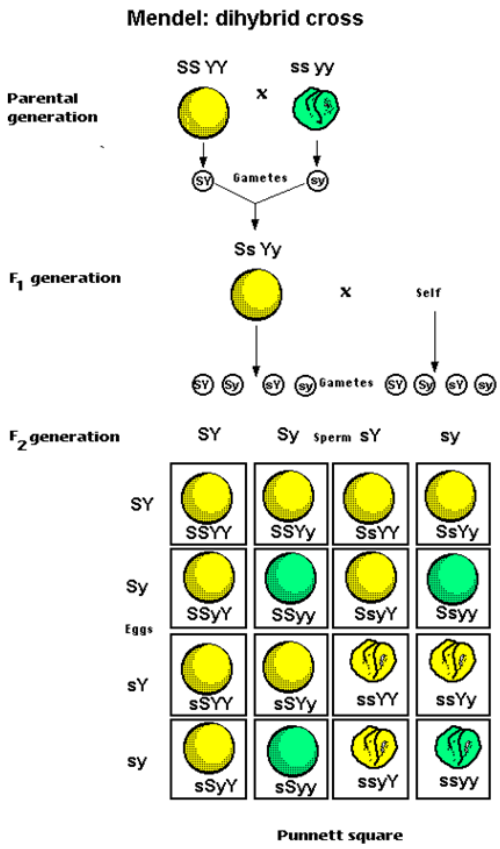
Independent Assortment

Alleles at the 2 gene loci segregate independently, and are NOT transmitted as a unit. Therefore, each plant would produce gametes with allele combinations that were not present in the gametes inherited from its parents.



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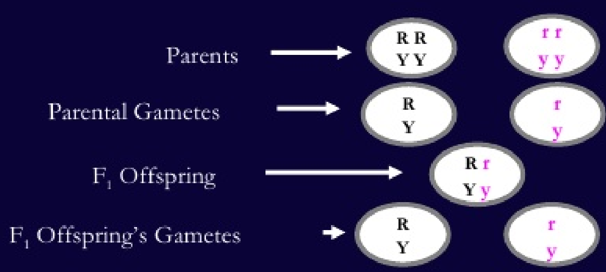
Dependent Segregation

- If dependent segregation (assortment) occurs:
  - Alleles at the 2 gene loci segregate together, and are transmitted as a unit.
  - Therefore, each plant would only produce gametes with the same combinations of alleles present in the gametes inherited from its parents:



What is the expected phenotypic ratio for the F_2?

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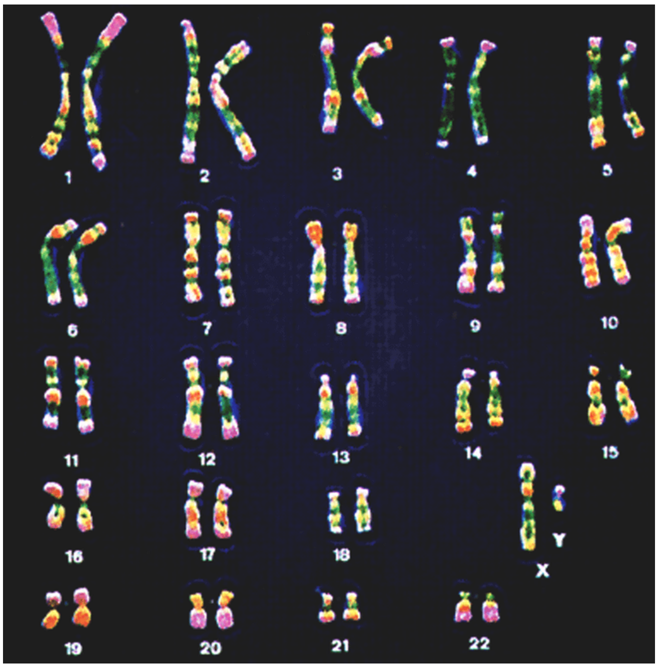
Chromosomes and Inheritance

Since genes are carried on chromosomes, knowledge of chromosome number and structure has far-reaching implications for basic genetics, human health, and evolution.



Normal human male karyotype

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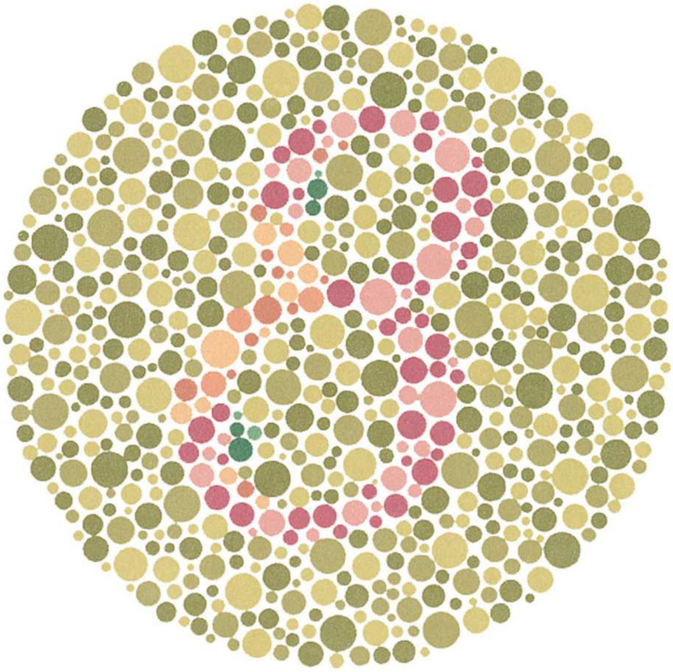
X-Linked Inheritance

Colorblindness is an X-Linked recessive trait



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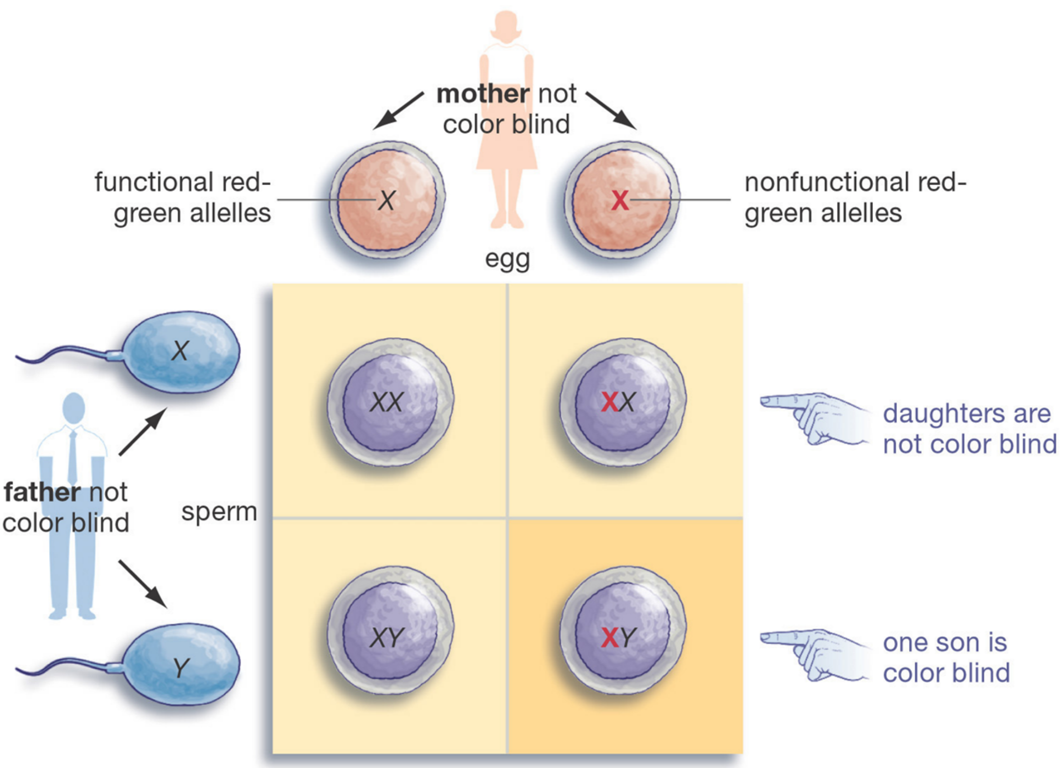

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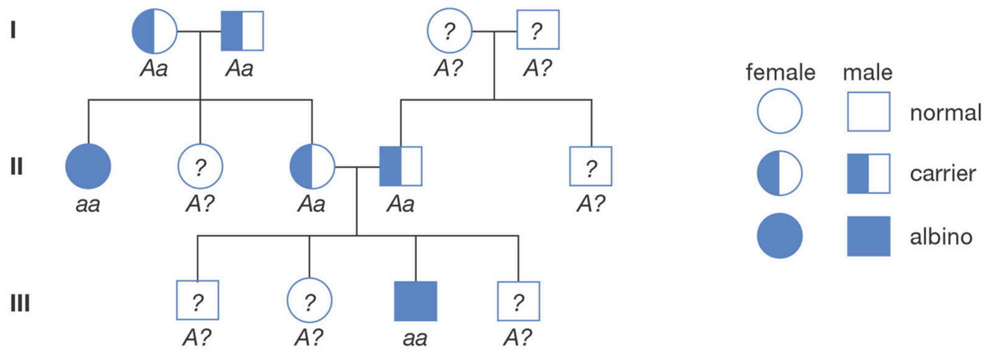
A pedigree analysis is a key tool in genetics!



Analyzing a pedigree is like puzzle-building – you try things (assigning potential genotypes) until the pieces fit (you're as certain as you can be about genotypes and modes of transmission (autosomal vs. X-linked; dominant vs. recessive).

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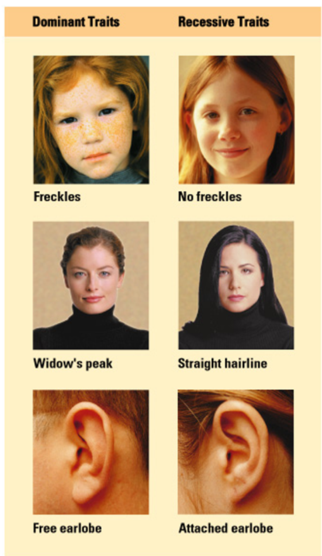
Pedigree Charts

As we have already discussed Mendel's findings can be applied to human traits as well.



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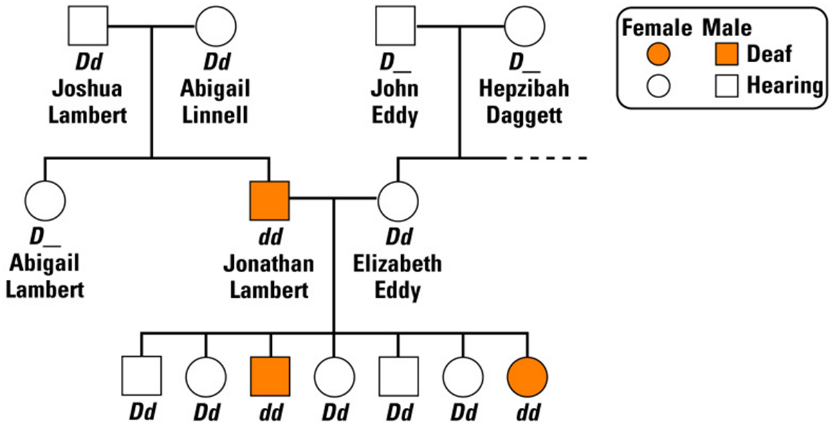
Family Pedigrees

- Shows the history of a trait in a family

- Allows researchers to analyze human traits



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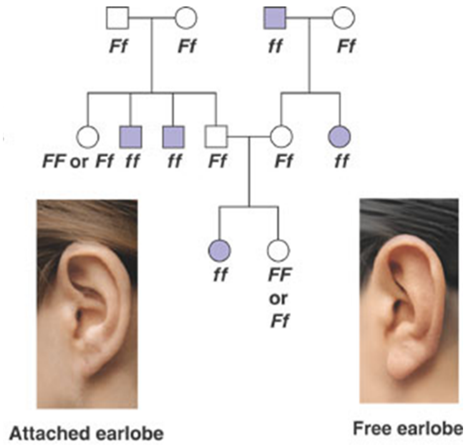
A Pedigree for a Recessive Trait



(b) Recessive trait (attached earlobe)

Those who have the gene but don't express it are termed carriers.

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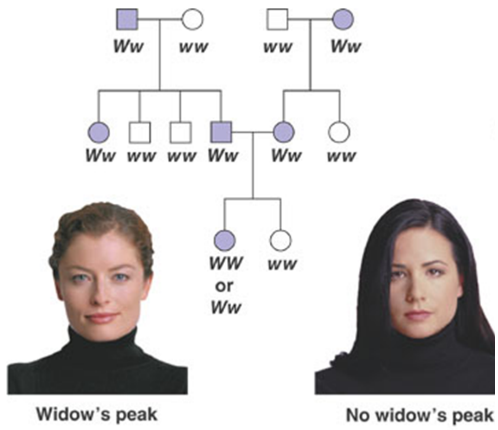
A Pedigree for a Dominant Trait



(a) Dominant trait (widow's peak)

Note that the trait appears in every generation and ½ the offspring of an affected heterozygote are expected to show the trait.

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Human Disorders Controlled by a Single Gene

- Many human traits:
  - Show simple inheritance patterns
  - Are controlled by genes on autosomes (not sex chromosomes)

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Table 9.1 Some Autosomal Disorders in Humans

Disorder	Major Symptoms	Incidence
Recessive disorders		
Albinism	Lack of pigment in skin, hair, and eyes	$\frac{1}{22,000}$
Cystic fibrosis	Excess mucus in lungs, digestive tract, liver; increased susceptibility to infections; death in infancy unless treated	$\frac{1}{1,800}$ European Americans
Galactosemia	Accumulation of galactose in tissues; mental retardation; eye and liver damage	$\frac{1}{100,000}$
Phenylketonuria (PKU)	Accumulation of phenylalanine in blood; lack of normal skin pigment; mental retardation unless treated	$\frac{1}{10,000}$ in U.S. and Europe
Sickle-cell disease (homozygous)	Sickled red blood cells; damage to many tissues	$\frac{1}{500}$ African Americans
Tay-Sachs disease	Lipid accumulation in brain cells; mental deficiency; blindness; death in childhood	$\frac{1}{3,500}$ Ashkenazi Jews
Dominant disorders		
Achondroplasia	Dwarfism	$\frac{1}{25,000}$
Alzheimer's disease (one type)	Mental deterioration; usually strikes late in life	Not known
Huntington's disease	Mental deterioration and uncontrollable movements; strikes in middle age	$\frac{1}{25,000}$
Hypercholesterolemia	Excess cholesterol in blood; heart disease	$\frac{1}{500}$ is heterozygous

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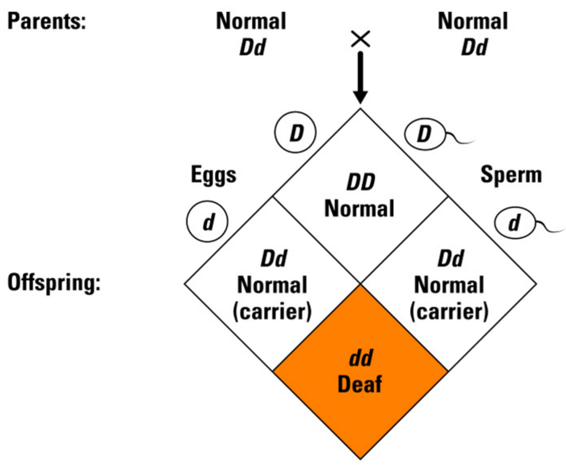
Recessive Disorders

- Most human genetic disorders are recessive
- Individuals can be carriers of these diseases



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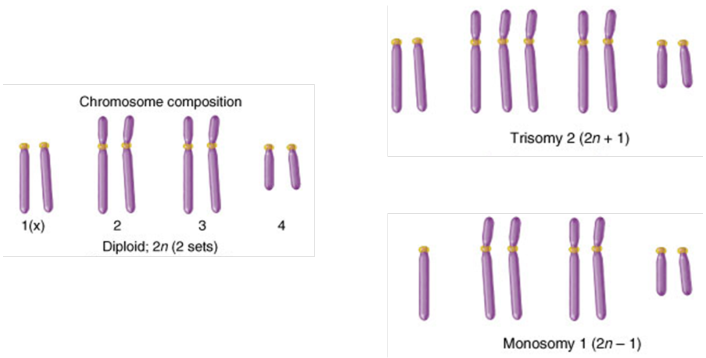
Changes in chromosome number and structure are important for health and evolution.

Ex. Aneuploidy - occurs when on of the chromosomes is present in an abnormal number of copies



Trisomy and monosomy are two types of aneuploidy

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Down Syndrome is caused by trisomy for chromosome 21.

Aneuploidy is remarkably common, causing termination of at least 25% of human conceptions.

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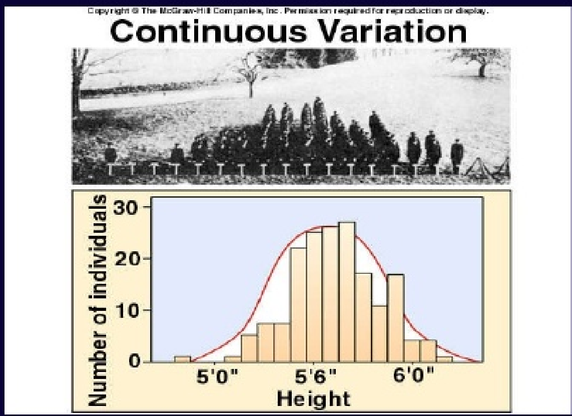
Polygenic Traits

- Most traits are not controlled by a single gene locus, but by the combined interaction of many gene loci. These are called polygenic traits.
- Polygenic traits often show continuous variation, rather then a few discrete forms:



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Epistasis

- Type of polygenic inheritance where the alleles at one gene locus can hide or prevent the expression of alleles at a second gene locus.
- Labrador retrievers one gene locus affects coat color by controlling how densely the pigment eumelanin is deposited in the fur.
- A dominant allele (B) produces a black coat while the recessive allele (b) produces a brown coat
- However, a second gene locus controls whether any eumelanin at all is deposited in the fur. Dogs that are homozygous recessive at this locus (ee) will have yellow fur no matter which alleles are at the first locus:

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Epistasis

ee
No dark pigment in fur

*E_*
Dark pigment in fur

eebb	*eeB_*	*E_bb*	*E_B_*
Yellow fur	Yellow fur	Brown fur	Black fur

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Pleiotropy

- This is when a single gene locus affects more than one trait.
- For example, in Labrador retrievers the gene locus that controls how dark the pigment in the hair will be also affects the color of the nose, lips, and eye rims.

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Environmental Effects on Gene Expression

- The phenotype of an organism depends not only on which genes it has (genotype), but also on the environment under which it develops.

- Although scientists agree that phenotype depends on a complex interaction between genotype and environment, there is a lot of debate and controversy about the relative importance of these 2 factors, particularly for complex human traits.

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